Francis S. Collins—Human Genome Project

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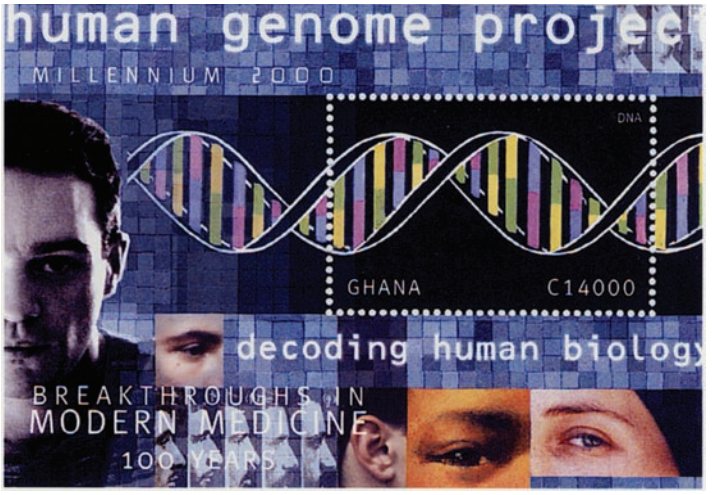

The American physician-geneticist Francis Sellers Collins became the second director of the National Center for Human Genome Research in April 1993. The position had been vacated the year before by James D. Watson (1928-), the Nobel laureate who codiscovered the chemical structure of DNA in 1953. As director of the Human Genome Project, Collins oversees a 15-year endeavor to completely map and sequence human DNA by 2005. Many scientists and medical researchers consider this project the most important scientific undertaking of our time. The work of the publicly funded Human Genome Project led by Collins complemented the work of the privately funded Human Genome Project led by J. Craig Venter (1946-) of Celera Genomics (Rockville, MD). Their cooperative efforts led to the simultaneous reporting of their data (Collins' work in *Nature*, February 15, 2001, and Venter's work in *Science*, February 16, 2001). Sequencing of the human genome represents the beginning of a new approach in biology and a new way of analyzing human disease. In 2000, the African country of Ghana issued a souvenir stamp (Scott No. 2215) that called attention to the Human Genome Project. This issuance provides an opportunity to honor Dr Francis S. Collins.

The youngest of 4 sons, Collins was born on April 14, 1950, in Staunton in north central Virginia, about 35 miles northwest of Charlottesville. His father had a PhD degree in English, taught at a local college, and raised livestock on the family farm. Collins was home-schooled by his mother until he entered 6th grade. While in high school, Collins became interested in chemistry. In 1966, at the age of 16 years, he entered the University of Virginia at Charlottesville and received a BS degree in chemistry in 1970. After receiving his baccalaureate degree, Collins enrolled at Yale University in New Haven, CT and received a PhD degree in physical chemistry in 1974. Collins was encouraged by Walter Gilbert (1932-), a future (1980) Nobel laureate in chemistry who had abandoned a promising career as a physicist to concentrate on molecular biology. In 1974, Collins entered medical school at the University of North Carolina in Chapel Hill. During his last year of medical school, Collins was introduced to the field of human genetics. In 1977, he received his medical degree from the university.

After receiving his MD degree, Collins completed both an internship and a residency at North Carolina Memorial Hospital in Chapel Hill. For the next 3 years, he was a fellow in human genetics and pediatrics at Yale University School of Medicine. In 1984, Collins left New Haven to join the staff of the University of Michigan Medical School at Ann Arbor as an assistant professor of internal medicine and human genetics. In 1987, he became chief of the division of medical genetics in the department of internal medicine, serving from 1987 to 1991. In 1988, he was promoted to associate professor of internal medicine and human genetics; in 1991, he became professor of those disciplines. During these years, Collins performed research, first as an assistant investigator (1987-1988), then

as an associate investigator (1988-1991), and finally as an investigator (1991-1993) with the Howard Hughes Medical Institute. In his research, Collins worked on what he called "positional cloning," a technique that has become a powerful component of modern molecular genetics because it allows identification of disease genes for almost any pathologic condition without previous knowledge of what might be the functional abnormality.

Using positional cloning techniques and working with Lap-Chee Tsui (1950-) and his colleagues at the Hospital for Sick Children in Toronto, Canada, Collins and his research team identified the gene for cystic fibrosis in 1989, the project's first major discovery. This discovery was followed by identification of the gene for neurofibromatosis in 1990.

In 1993, a successful joint effort with the Huntington's Disease Research Group, which comprised laboratories in the United States, England, and Wales, led to the discovery of the gene that causes Huntington disease. In addition, success has been achieved in identifying the gene for multiple endocrine neoplasia type 1. Moreover, Collins' laboratory is also trying to identify the genetic basis of type 2 diabetes mellitus by studying a large cohort of affected sibling pairs and relatives in Finland. The project involves a search for genes that confer susceptibility to diabetes or intermediate traits such as insulin resistance in more than 5000 persons. A genome scan followed by fine mapping has identified regions of chromosomes 20, 22, 11, 6, X, and 3 that apparently harbor the diabetes susceptibility genes. High-throughput genotyping of single nucleotide polymorphisms using mass spectrometric technology is also being used in search of the precise variant responsible.

Francis S. Collins has been recognized for his achievements by being elected to the Institute of Medicine and the National Academy of Sciences (1993). He has received numerous national and international awards, including the Gairdner Foundation International Award for his work on cystic fibrosis (1990). He became Director of the National Institutes of Health in 2009.